Indian Journal of Pediatrics (August 2022) 89(8):818
https://doi.org/10.1007/s12098-022-04239-4

SCIENTIFIC LETTER

Siblings with Proximal Muscle Weakness—Rigid Spine Is the Clue!

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Received: 3 March 2022 / Accepted: 7 April 2022 / Published online: 14 June 2022

To the Editor: A 9-y-old girl born to a nonconsanguineous couple presented with complaints of difficulty in running and climbing stairs. She also needed support to stand from sitting position. The child could walk with a waddling gait. Other developmental domains were age-appropriate. The elder sibling (male) had a similar history with severe involvement and succumbed at 5 y of age. On examination, difficulty in getting up from supine position (truncal weakness +), symmetrical proximal predominant muscle weakness, and Gower sign were observed. Restriction in the cervical spine mobility was noticed. DTRs were elicitable. There was no calf hypertrophy, myotonia, or polyminimyoclonus. The possibility of rigid spine muscular dystrophy was entertained. The child had normal creatine phosphokinase levels on investigation, and the clinical exome revealed a homozygous nonsense mutation in the exon 9 of the *SEPN1* gene c.1180G>T (p. Glu394Ter).

Rigid spine syndrome is characterized by rigidity of the spine secondary to contractures in extensors and weakness in spinal flexors. The stiffness of the spine can be a later manifestation of any of the neuromuscular disorders, which affect the spinal flexor group of muscles [1, 2]. Rigid spine muscular dystrophy (RSMD) is an autosomal recessive condition, presenting with early-onset, severe neck stiffness. RSMD predominantly affects the axial muscles with early respiratory and minimal cardiac involvement, whereas rigid spine associated with other myopathies predominantly involves proximal muscles and early cardiac involvement and has neck stiffness later in the disease course [3, 4].

Whenever we encounter a child with suspicion of neuromuscular disorder, it is prudent to examine all groups of muscles involved for narrowing down the differentials. In our case inability to flex the neck provided us with an important hint. A meticulous clinical examination involving all the groups of muscles can reveal various diagnostic clinical signs, which avoid unnecessary investigations in children with neuromuscular ailments.

Declarations

Conflict of Interest None.

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